Patient Name: ARUPTEST, BCG GAA 1

Date of Birth: 10-Sep-10 Gender: Male ARUP ID: 10003

Requisition #: Client Supplied ID:

> Physician: ARUP, ARUP Printed: 23-Jun-17 09:24:27

ARUP Example Report Only 004848 500 Chipeta Way Salt Lake City UT 84108

							Reported/
Procedure	Result	<u>Units</u>	Ref Interval	Accession	Collected	Received	Verified
Pompe GAA Activity Leukocytes Interp	AAA Activity Leukocytes Interp See Note f			17-174-900022	23-Jun-17	23-Jun-17	23-Jun-17
					08:44:00	08:44:00	09:22:46
Pompe GAA Activity Leukocytes	<0.4 L	nmol/h/mg	[5.5-25.0]	17-174-900022	23-Jun-17	23-Jun-17	23-Jun-17
					08:44:00	08:44:00	09:22:46

23-Jun-17 08:44:00 Pompe GAA Activity Leukocytes Interp:

In this sample, lysosomal acid alpha-glucosidase (GAA) activity was undetectable, and consistent with a diagnosis of Pompe disease (OMIM#606800, Glycogen Storage Disease II). This test cannot predict disease severity. Additionally, this test does NOT evaluate enzyme activities deficient in other lysosomal storage disorders. Consideration should be given to DNA testing to confirm this finding. Genetic evaluation is recommended.

23-Jun-17 08:44:00 Pompe GAA Activity Leukocytes Interp:

INTERPRETIVE INFORMATION: Pompe Disease (GAA), Enzyme

Activity in Leukocytes

Units of measure for Pompe Disease (GAA) Activity are: Nanomoles of substrate hydrolyzed per hour per mg of protein.

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Chart ID: 12272162

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